

# Newborn Screening Update

## Michigan Newborn Screening Program

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## INTRODUCING .....

Beginning with this issue, each issue will include an introduction of a member of the Michigan Newborn Screening staff and their colleagues..

Midge McCaustland, RNC, MSN, joined the Program as the Nurse Consultant/Newborn Screening Educator on January 3, 2005. Midge has a broad range of experience in Maternal Child Nursing. In addition to various OB nursing positions, Midge was a Clinical Nurse Specialist/Specialized Care Manager in the Specialized Home Care Program/Children's Special Health Care Services. In this position, she helped coordinate the transition of special needs children from the hospital to the home. Most recently, Midge was a Nurse Consultant with the Michigan Department of Community Health's Division of Licensing and Certification, conducting surveys of various health care facilities to ensure that the facility was in compliance with both state and federal rules and regulations.

Midge will be contacting all birthing hospitals and hospitals with an NICU to schedule a visit to discuss their Newborn Screening practices. In addition, Midge is available to present various Newborn Screening topics and educational materials. She can be reached at 517-335-8588 or [mccaustlandm@michigan.gov](mailto:mccaustlandm@michigan.gov).

## What happened to the PKU test???



It expanded and grew and is now referred to as ***the Newborn Screening (NBS) test***. Those of us who are seasoned practitioners can remember 1965 when we were first instructed to obtain that "big" drop of blood from that "tiny" heel. At that time, babies were screened for Phenylketonuria (PKU) so that, if the disorder was detected early, treatment could be initiated. Since then, advancements in technology have enabled the early detection of more disorders from one newborn screen. In Michigan, we screen for 11 disorders including PKU. The others that were added over time are: Congenital Hypothyroidism [CH] (1977), Galactosemia (1984), Maple Syrup Urine Disease [MSUD], Biotinidase Deficiency, Hemoglobinopathies (1987), Congenital Adrenal Hyperplasia [CAH] (1993), Medium Chain Acyl CoA Dehydrogenase Deficiency [MCAD] (2003), and Argininosuccinic Aciduria [ASA], Citrullinemia, and Homocystinuria (2004). So, please remember to use the correct terminology when talking to families and other individuals.

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**IMPORTANT!!!**  
**See page 2 for information on the pink NBS repeat screen card.**

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## A Few Reminders ...

### **REPEAT SAMPLE EXPIRATION DATE DISCREPANCY**

The **REPEAT SAMPLE** pink newborn screening (NBS) cards, **with the preprinted** orange Biohazard label on the flap covering the blood spots, **expire at the end of December 2005**. This correct expiration date is printed on the filter paper near the circles. The top pink form and the yellow submitter copy have an incorrect expiration date of March 2005. Please be assured that the expiration date on the filter paper is correctly labeled.

The repeat sample pink newborn screening cards **without the preprinted** orange biohazard label on the flap **do expire** at the end of March 2005. If necessary, Contact Valerie Klasko at 517-241-5583 to order current REPEAT SAMPLE newborn screening cards to replace these cards before they expire.

For questions or concerns regarding newborn screening please contact, Midge McCaustland, NBS Educator at 517-335-8588.

### **Reviewing the information on the NBS card with the mother**

Before the blood is collected the information on the NBS card needs to be verified with the mother. This includes her name, address, phone, social security number and the name and address of the health care provider who will be caring for the infant after discharge from the hospital. This is also an ideal time to provide the mother with the *Newborn Screening brochure*.

### **Physician Responsible for Care of the Newborn**

Please be sure that the physician listed on the NBS card is the physician who will be caring for the newborn after discharge from the hospital. This physician will be notified of unsatisfactory or presumptive positive newborn screening results and is responsible for all retests. If the mother does not identify a health care provider for her infant, the hospital should identify a designated person who will be responsible for the prompt follow-up of unsatisfactory and presumptive positive results. **It is the submitter's responsibility to ensure that all information on the NBS card is correct.**

### **What causes a delay in reporting results?**

Incomplete information on the NBS card can result in delays in reporting results. It is extremely important to ensure that the *infant's birth weight, date and time of birth and the date and time the sample was obtained* are accurately recorded in the designated spaces on the NBS card. **It is the submitter's responsibility to ensure that all information on the NBS card is correct.** Help us report the results promptly!

### **Newborn Screening Results**

The Newborn Screening Laboratory sends two copies of screening results. One for the submitter to keep, and one to forward to the physician listed on the screening card. **It is the submitter's responsibility to forward screening results to the physician listed on the card.**

### **Newborn Screening Education Program**

Just a reminder that the Newborn Screening Educational Online Tutorial is accessible from the NBS website [www.michigan.gov/newbornscreening](http://www.michigan.gov/newbornscreening). Click on the link and follow the directions to register for the course , *free of charge*.

# A MOTHER'S STORY ...

When I was asked to write an article about my daughter, I reflected back on how she has changed our lives. Bethany is 4 ½ yrs old and she has Classic PKU. When she was born she had the routine Newborn Screening in the hospital. We didn't think anything of it. I had worked as an OB nurse and had poked many little heels without a second thought. These conditions were rare and you never heard of anyone diagnosed with any of them. We received a call that we needed to take Bethany back to the OB floor for a repeat screening. I had a letter with the Metabolic Clinic address on it and when I showed it to the clerk she told us "We don't need that, we send them out all the time". We were reassured that there were many "false positives" and we shouldn't worry. We then received a second call from the OB floor to come back for a second repeat. I was concerned and only when we went back in did I find out that the first repeat was sent to the state lab and not to the Metabolic Clinic for a positive diagnosis, wasting precious time.

Bethany was diagnosed when she was about 8 days old. PKU?! What is the treatment? Will she be normal? Whew? We were devastated to say the least. We were then contacted by the dieticians at the Clinic who told me to stop breastfeeding. Breast milk has about 50mg of phenylalanine per oz. We were told we would need to start her on formula and that same day we received the formula. We were allowed to restart the breastmilk in small amounts after our first Clinic visit the next week. We were encouraged to attend the winter party for kids with metabolic conditions. Bethany was 5 months old and what a relief it was for us to see PKU kids running, jumping, laughing and yes-whining just like any other kids. These kids didn't have the speech or gait problems, or the musty smell or any other complications of untreated PKU.

Dietary changes were and still are done almost weekly as her tolerance for "phe" changes. Adding baby food at 6 months was stressful- we scraped food off the floor, the dogs and out of hair to reweigh it on the gram scale in an attempt to get an accurate phe intake for the day. We have been able to maintain levels within the recommended 2-6mg/dl almost consistently. She will drink her modified protein formula for the rest of her life to obtain her essential amino acids. We work with Bethany and her diet at every opportunity-when we go out to eat we pack hers, when we travel we pack hers, when we cook we talk about foods and what amounts she can have.

The emotions are still there-like going to a party and watching the other 11 kids lick the frosting off their fingers only to find out that is cream cheese frosting and made with milk and is too high in phe. We have also worked with her daycare provider and will eventually work with her school.

I look at this beautiful little girl with her blond curly hair and blue eyes and am proud that she has reached every milestone and excelled at many. She is able to count to over 60, ice skates, does gymnastics and is in preschool. Once I asked her what PKU was, she told me it was about not eating some foods. She told me she has PKU and when I asked her if she can drink mommy and daddy's milk she said "no it not good for my body". She tells us that her milk comes from the blender! She is simply amazing in all that she knows already. When I think ahead to all the challenges she will face- her teen years and going out with peers, her college days and the cafeteria, and eventually having her own family she will cook for, I can say that we have provided the foundation for her diet and hopefully she will make great choices as she becomes more independent. We are very proud of her. Without the Newborn Screening program and her being diagnosed, Bethany would face losing brain cells and the possibility of mental retardation.

Submitted by: Sandy LaPrad, Mother of Bethany, child diagnosed with Classic PKU

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**Michigan Newborn Screening  
Program**

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